

Lack of association of C282Y and H63D mutations in the hemochromatosis (HFE) gene with diabetes mellitus type 2 in a case-control study of women in Brazil

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ABSTRACT. Hereditary hemochromatosis is one of the most common autosomal recessive diseases; it is characterized by excess absorption of iron. Clinically, the major challenge is to diagnose increased iron deposition before irreversible tissue damage has occurred. C282Y and H63D are the main mutations related to hereditary hemochromatosis; these mutations have been reported to be associated with increased risk of developing diabetes mellitus type 2 (DM2). We investigated whether these mutations are associated with increased risk for the development of DM2 in women in Brazil. Seventy-two women with clinical diagnosis of DM2 under treatment with hypoglycemic agents and a control group composed of 72 women with no clinical history of diabetes were studied. The C282Y and H63D mutations were determined by PCR-RFLP. Significant differences were not observed for C282Y and H63D, when we compared diabetic and non-diabetic women. We suggest that mutations C282Y and H63D in the HFE

gene are not significant risk factors for the development of DM2 in Brazilian women.

Key words: Hereditary hemochromatosis; HFE; C282Y; H63D; Diabetes mellitus type 2; Women